



Primitive Neuroectodermal Tumour: A Rare Association with Neurofibromatosis Type 1

Primitive Nöroektodermal Tümör: Nörofibromatosis Tip 1 ile Nadir Birliktelik

Primitive Nöroektodermal Tümör / Primitive Neuroectodermal Tumour

Hatice Gümüş¹, Metehan Gümüş², Uğur Fırat³, Hakan Önder⁴, Fatih İnci⁵,

¹Department of Radiology, ²Department of General Surgery, ³Department of Pathology,

⁴Department of Radiology, Medical School, Dicle University, Diyarbakir, ⁵Department of Radiology, Elazığ Harput State Hospital, Elazığ, Turkey

Özet

Nörofibromatosis tip 1 (NF-1), benin tümörleri içeren yaygın herediter bir hastalıktır. Omuriliğin primitive nöroektodermal tümörü (PNET) oldukça nadirdir. Biz burada NF-1'li bir kadın hastada paravertebral alandaki spinal epidural PNET'in US, BT ve MRG bulgularını sunuyoruz. 37 yaşındaki NF-1'li kadın hastanın bel ağrısı ve sağ alt ekstremitede ilerleyici güçsüzlüğü mevcuttu. US'da batında heterojen iyi sınırlı kitle lezyon izlendi. BT ve MRG'de L5-S1 düzeyinde sağ nöral foramen kaynaklı heterojen kontrastlanan kitle lezyon mevcuttu. Lezyon komşuluğunda posterolateral karın duvarında mezenter ve barsak anslarının bulunduğu büyük bir fıtık kesesi vardı. Tanı hastaya transabdominal tru-cut biyopsi yapılarak konuldu. Nadir olmasına rağmen, spinal epidural lezyonlarda PNET ihtimali akıldan tutulması gerekmektedir.

Anahtar Kelimeler

Spinal Tümör; Primitive Nöroektodermal Tümör; Nörofibromatosis Tip 1

Abstract

Neurofibromatosis type 1 (NF-1), is a common hereditary disease which often take the form of benign tumours. Spinal primitive neuroectodermal tumour (PNET) is very rare. Herein we report US, CT and MRI findings of a female patient with PNET involving the spinal epidural and paravertebral spaces with neurofibromatosis type 1. A 37-year-old woman has NF-1 presented with lower back pain and progressive weakness of the right lower extremities. US revealed a heterogenous well defined solid mass. CT and MRI revealed heterogeneously enhanced mass originating from the right neural foramen at the level of L5-S1. Adjacent to the lesion, there was a large hernia sac, including bowel loops and mesentery, in the posterolateral abdominal wall. The definitive diagnosis was made by transabdominal percutaneous tru-cut biopsy. Although it is rare, the possibility of PNET should be kept in mind in assessing the spinal epidural lesions.

Keywords

Spinal Tumour; Primitive Neuroectodermal Tumour; Neurofibromatosis Type 1

DOI: 10.4328/JCAM.867

Received: 05.12.2011 Accepted: 19.01.2012

Printed: 01.01.2015

J Clin Anal Med 2015;6(1): 114-6

Corresponding Author: Hatice Gümüş, Department of Radiology, Medical School, Dicle University, Yenişehir 21280 Diyarbakir, Turkey.

T.: +90 4122488 58 F.: +90 4122488115 E-Mail: drhaticegumus@hotmail.com

Introduction

Neurofibromatosis type 1 (NF-1), is a common hereditary disease in which the skin, nervous system, bones, endocrine glands, and sometimes other organs are the sites of a variety of congenital abnormalities, which often take the form of benign tumors [1]. Spinal primitive neuroectodermal tumor (PNET) is very rare. Thirty-one patients have been reported with such tumors [2], and most of the tumors were intramedullary; only 7 patients had tumors located epidurally. PNETs are aggressive neoplasias that usually diagnosed in infancy. PNET in adulthood is rare and, exceptionally, in association with neurofibromatosis type 1 (NF-1) [3]. Bohn Sarmiento et al. [3] reported a patient with neurofibromatosis type 1 who had lumbar region intra-spinal PNET. To our knowledge there are a few cases of PNET combined with neurofibromatosis type 1 in the English literature [3, 4, 5].

Case Report

A 37-year-old woman presented with lower back pain projecting down to the right leg with progressive weakness of the right lower extremities. The patient with multiple cafe-au-lait spots and neurofibromas in the skin had been diagnosed neurofibromatosis type 1 previously. Transabdominal US revealed a heterogenous well defined solid mass with central necrosis displacing iliac veins laterally in the right lower quadrant. In the right posterolateral adjacent to the mass, there was a large hernia sac including bowel loops and mesenteric vascular structures (Fig. 1).

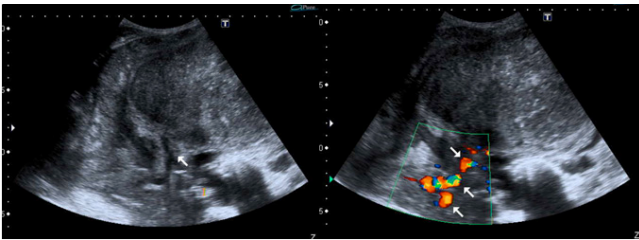


Figure 1. Abdominal US. US show a large hernia sac including bowel loops and mesenteric vascular structures in the right posterolateral of the mass

CT revealed bilateral meningocele at the level of L5-S1. At this level, a heterogeneous necrotic centered mass with well-defined borders was originating from the right neural foramen. The large, hypodense, heterogeneously enhanced mass caused vertebral scalloping, and displaced bowels and vascular structures laterally. Adjacent to the lesion, there was a large hernia sac, including bowel loops and mesentery, in the posterolateral abdominal wall (Fig. 2). The mass was hypointense on T1W, hyperintense on T2W, and enhanced heterogeneously on MRI (Fig. 3). Cranial MRI was normal.

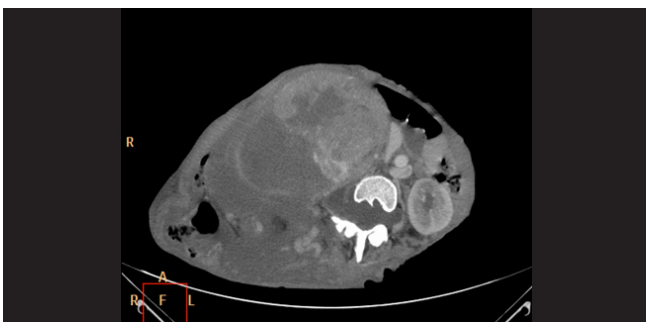


Figure 2. Axial abdominal CT. There is bilateral meningocele at the level of L5-S1 and vertebral scalloping

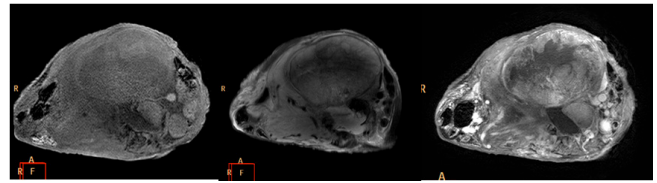


Figure 3. T1, T2 and postcontrast T1-W axial MR images. The lesion show hypointens on T1 W, hyperintens on T2-W and enhanced heterogeneously on postcontrast T1-W images

The patient had transabdominal percutaneous tru-cut biopsy. Microscopic examination revealed that the tumor was composed of undifferentiated small, round to oval appearing cells with nuclei containing fine chromatin, scanty eosinophilic cytoplasm, and indistinct cytoplasmic membrane producing a syncytial appearance (Fig. 4). And a few mitoses were seen on per 10 high-power-fields. The tumor cells were immunohistochemically positive for CD99 (MIC2) (Fig. 5), Neuron Specific Enolase (NSE), Vimentin, S-100 and CD68. Other immunohistochemical markers including Chromogranine, Synaptophysin, Melan-A, EMA and Pancytokeratin (PanCK) were negative. The patient was referred to Oncology for radiation and chemotherapy.

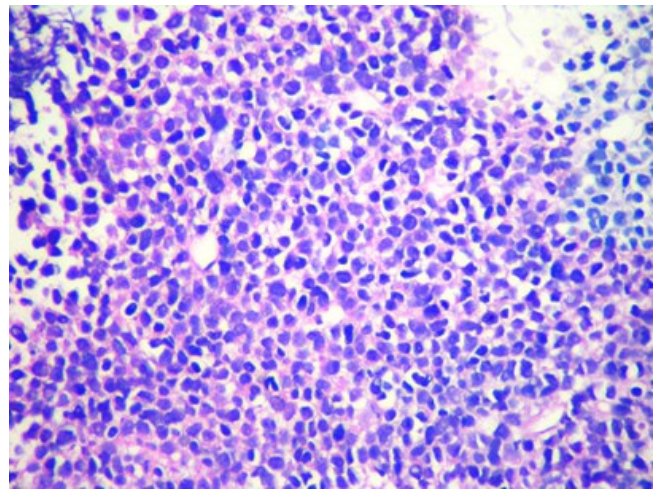


Figure 4. The tumor showing small round to oval appearing atypical cells with indistinct cytoplasmic membrane (H&E stain, X200)

Discussion

Ewing's sarcoma /PNET arising from the lumbosacral spine is reported very rarely. PNET needs to be differentiated from other tumors (schwannoma, neurofibroma, lymphoma, malignant peripheral nerve sheath tumors (MPNST)) which are arising in

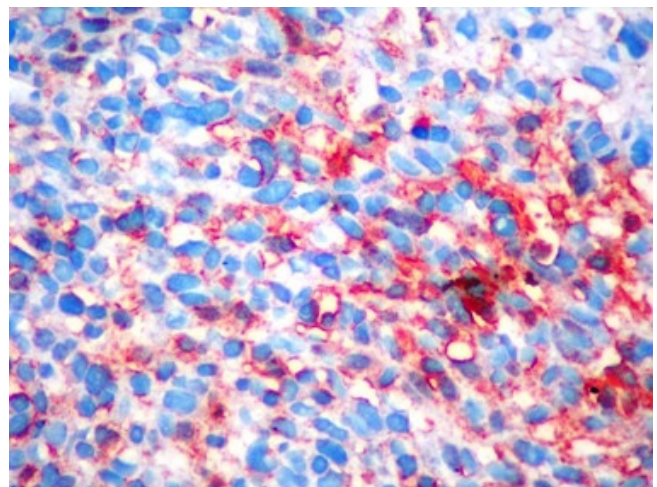


Figure 5. CD99 positivity in the tumor cells (Immunoperoxidase stain, X400)

this region [6]. The most useful diagnostic imaging methods are computerized tomography (CT) and magnetic resonance imaging (MRI) [7]. PNET may exhibit a well defined hypodense mass displacing the thecal sac on nonenhanced CT scans, and heterogeneous contrast enhancement on contrast-enhanced CT scans. Central, nonenhancing areas show necrosis within the tumor. MRI is useful for determining the extent of the tumor because of its high soft tissue resolution. The tumour is usually hypointense to the spinal cord on T1-W images and hyperintense on T2-W images. Mild to moderate and homogeneous or heterogeneous enhancement can be seen on postcontrast MRI [7]. Schwannoma is the most common intradural extramedullary neurogenic tumor in the spine. Schwannomas are well-encapsulated, compressing the cord, conus to the contralateral side. These tumors have a cystic component (40% cases), and show peripheral enhancement on postcontrast T1W images [8]. Neurofibromas are well-defined lesions. The tumors show a hyperintense rim and a hypointense center on T2-W and postcontrast T1-W images. They are also often associated with NF-1 [6]. Schwannoma and neurofibroma were considered in our patient because of CT and MRI characteristics of lesions.

Lymphomas (non-Hodgkin's lymphoma) present in older age-groups and spread via the subarachnoid space, causing diffuse thickening of the spinal nerve roots. They show marked enhancement on postcontrast MRI images [6]. Lymphoma was not considered in our patient because she was 37 year old.

Approximately 5% of patients with NF-1 develop MPNST, which usually arises from a plexiform neurofibroma [9]. MPNSTs are usually infiltrative and heterogenous, and have irregular margins on CT and MRI. These tumours have central hemorrhage and necrosis. They show variable patterns of enhancement, and they commonly show diffuse or peripheral heterogeneous contrast enhancement on postcontrast CT and MRI [10]. MPSNT was not considered in our patient because CT and MRI showed well-defined lesions.

In this case, we established our radiological diagnosis as benign nerve sheath tumor (schwannoma, neurofibroma) in the patient with NF-1. Because of the rare occurrence, spinal PNET was not considered before the histologic examination. Diagnosis could be established only after the biopsy. Differentiation of these rare lesions requires imaging studies along with histopathological confirmation [6].

Spinal epidural PNET is a rare neoplasm that difficult to diagnose. PNET in association with NF-1 is very rare (there is only one other case in literature). CT and MRI seem to be effective in the overall assessment. Although it is rare, the possibility of PNET should be kept in mind in assessing the spinal epidural lesions.

Competing interests

The authors declare that they have no competing interests.

References

1. Fortman BJ, Kuszyk BS, Urban BA, Fishman EK. Neurofibromatosis Type 1: A Diagnostic Mimicker at CT1. *Radiographics* 2001;21(3):601-12.
2. Dorfmueller G, Wurtz FG, Umschaden HW, Kleinert U, Ambros PF. Intraspinale primitive neuroektodermale Tumoren: Report of two cases and review of the literature. *Acta Neurochir* 1999;141(11):1169-75.
3. Bohn SU, Aguiar BD, Camacho GR, Rivero VJC, Aguiar MJ. Lumbar region intraspinal primitive neuroektodermale Tumoren (PNET) combined with neurofibromato-

sis type 1. *Clin Transl Oncol* 2005;7(10):464-7.

4. Chan GC, Nicholls JM, Lee AC, Chan LC, Lau YL. Malignant peripheral Neuroectodermal Tumor in an Infant With Neurofibromatosis Type 1. *Med Pediatr Oncol* 1996;26(3):215-9.

5. Mulholland CB, Barkhoudarian G, Cornford ME, McBride DQ. Intraspinale primitive neuroektodermale Tumor in a man with neurofibromatosis type 1: Case report and review of the literature. *Surg Neurol Int* 2011;2:155.

6. Narula MK, Gupta N, Anand R, Kapoor S. Extraskeletale Ewing's Sacroma /primitive neuroektodermale Tumor of the sacral Nervenplexus. *Indian J Radiol Imaging* 2009;19(2):151-4.

7. Ozturk E, Mutlu H, Sonmez G, Aker FV, Basekim CC, Kizilkaya E. Spinal epidural extraskelale Ewing Sarkoma. *J Neuroradiol* 2007;34(1):63-7.

8. Kim SB, Kim HS, Jang JS, Lee SH. Mobilität von intradural extramedulläre Schwannoma an spine: Report of three cases with literature review. *J Korean Neurosurg Soc* 2010;47(1):64-7.

9. Kim JG, Sung WJ, Kim DH, Kim YH, Sohn SK, Lee KB. Malignant Peripherale Nerven Sheath Tumor in Neurofibromatosis Type 1: Unusual Presentation of Intraabdominale or Intrathoracale Mass. *Korean J Intern Med* 2005;20(1):100-4.

10. Murphey MD, Smith WS, Smith SE, Kransdorf MJ, Temple HT. From the Archives of the AFIP. Imaging of muskuloskeletale Neurogenische Tumoren: Radiologische-Pathologische Korrelation. *Radiographics* 1999;19(5):1253-80.